



NGLY1 gene

N-glycanase 1

Normal Function

The *NGLY1* gene provides instructions for making an enzyme called *N*-glycanase 1. This enzyme is involved in a process called deglycosylation, by which chains of sugar molecules (glycans) are removed from proteins. Specifically, *N*-glycanase 1 removes glycans from misfolded proteins. This step is thought to be essential for certain abnormal proteins to be broken down (degraded).

Health Conditions Related to Genetic Changes

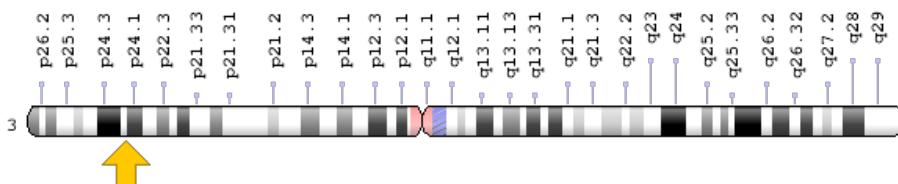
NGLY1-congenital disorder of deglycosylation

At least 13 mutations in the *NGLY1* gene have been found to cause *NGLY1*-congenital disorder of deglycosylation (*NGLY1*-CDDG). This condition affects many body systems, causing delayed development, movement abnormalities, problems with liver function, eye abnormalities, and a reduction or absence of tears (hypolacrima or alacrima). These mutations impair production of the *N*-glycanase 1 enzyme, resulting in a severe reduction or absence of the enzyme's function. Without the removal of glycans, certain misfolded proteins cannot be broken down. It is thought that the abnormal proteins accumulate and form clumps (aggregates) in cells. These aggregates may damage cells in the brain, liver, and eyes, leading to the signs and symptoms of *NGLY1*-CDDG.

Chromosomal Location

Cytogenetic Location: 3p24.2, which is the short (p) arm of chromosome 3 at position 24.2

Molecular Location: base pairs 25,718,944 to 25,790,039 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CDDG
- CDGIV
- FLJ11005
- hPNGase
- peptide-N(4)-(N-acetyl-beta-glucosaminyl)asparagine amidase isoform 1
- peptide:N-glycanase
- PNG1
- PNGase

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Improperly Folded Proteins Are Exported from the ER and Degraded in the Cytosol
https://www.ncbi.nlm.nih.gov/books/NBK26841/#_A2236_

Clinical Information from GeneReviews

- NGLY1-Related Congenital Disorder of Deglycosylation
<https://www.ncbi.nlm.nih.gov/books/NBK481554>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28NGLY1%5BTIAB%5D%29+OR+%28N-glycanase+1%5BTIAB%5D%29%29+OR+%28%28CDDG%5BTIAB%5D%29+OR+%28PNG1%5BTIAB%5D%29+OR+%28PNGase%5BTIAB%5D%29+OR+%28hPNGase%5BTIAB%5D%29+OR+%28N-glycanase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- N-GLYCANASE 1
<http://omim.org/entry/610661>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_NGLY1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NGLY1%5Bgene%5D>

- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:17646
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:55768>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/55768>
- UniProt
<https://www.uniprot.org/uniprot/Q96IV0>

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/NGLY1>

Reviewed: August 2017

Published: June 23, 2020

Lister Hill National Center for Biomedical Communications
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